



Pes Kavus ve Charcot Marie Tooth Hastalığı: Olgu Sunumu ve Literatürün Gözden Geçirilmesi

Pes Cavus and Charcot Marie Tooth Disease: A Case Report and Brief Review of the Literature

Pes Cavus and Charcot Marie Tooth Disease / Pes Kavus ve Charcot Marie Tooth Hastalığı

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Özet

Charcot-Marie-Tooth (CMT) hastalığı hem genetik hem de klinik olarak oldukça heterojen bir hastalıktır. Tanı için klinik ve elektrofizyolojik veriler esastır. CMT li çocuklar erken yaştan itibaren edinilmiş ayak zayıflığı, kontraktür ve deformiteler (pes kavus ve çekiç parmaklar) geliştirebilirler. CMT de ayak ve ayak bileğini hedef alan erken girişimler uzun dönem özürüllüğü önleyebilir. Burada biz sonradan edinilmiş pes kavuslu ve çekiç parmaklı bir CMT hastasını sunmakta ve kısaca CMT nin tanısı, tedavisi ve rehabilitasyonu için literatür taraması yapmaktayız. Sonuç olarak biz sonradan edinilmiş pes kavus ve çekiç parmağın ayırıcı tanısında CMT nin de akla gelmesi kanısındayız.

Anahtar Kelimeler

Pes Kavus, Çekiç Parmak, Charcot Marie Tooth Hastalığı.

Abstract

Charcot-Marie-Tooth (CMT) is a disease that is highly heterogeneous, both clinically and genetically. Clinical and electrophysiological data are essential for diagnosis. Children with CMT experience acquired foot weakness, contracture and deformity (pes cavus and hammer toes) from an early age. Early intervention targeting the foot and ankle may prevent long-term disability in CMT. Here we present a CMT patient with acquired pes cavus and hammer toes and review the literature briefly for diagnosis, treatment, and rehabilitation of CMT. As a result we conclude that CMT should also come into mind in the differential diagnosis of acquired pes cavus and hammer toes.

Keywords

Pes Cavus, Hammer Toe, Charcot Marie Tooth Disease.

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Introduction

The hereditary motor and sensory neuropathies (HMSN) also called Charcot-Marie-Tooth disease (CMT) describe a spectrum of slowly progressive inherited disorders affecting peripheral motor and sensory nerves. The underlying pathophysiology is a specific mutation in one of several myelin genes that results in compromise of the myelin integrity [1]. It is classified according to clinical features, genetic inheritance, and electrophysiological and histopathological findings. The Dyck classification developed in the 1970s subdivided the HMSN (CMT) into types 1 through 7 based upon the clinical and electrophysiologic features [2]. The prevalence of CMT ranges from a rate of 8–41 per 100,000 people depending on the geographical region of origin [3]. The most common subtypes are CMT1 (demyelinating with autosomal dominant transmission) and CMT2 (axonal and usually dominant). CMT1A is the most common subgroup which results from the duplication of a genomic fragment that encompasses the PMP22 gene on chromosome 17 [4]. CMT1A affects 60 to 80% of the general CMT population [5]. The wide range of foot/ankle manifestations in CMT1A complicates the assessment, diagnosis and therapy. Children with CMT1A experience foot weakness, contracture and deformity, pes cavus from an early age [6]. The disease is characterized by weakness and atrophy of the distal muscle groups of the limbs, gait disturbance, impaired sensation in the hands and feet, decreased and/or absent deep tendon reflexes, and skeletal deformities such as pes cavus and hammer toes. Sensory loss of all modalities is seen [7]. Pes cavus is thought to be an almost universal manifestation of CMT1A, particularly in older children and adults [6,8]. In previous studies, pes cavus was apparent in early childhood in only 11-33 % of cases, but it increased to 62.5-80% during adolescence [7,9]. Foot and ankle muscle imbalance, reduced strength in all muscles of feet, intrinsic foot weakness, restriction of ankle dorsiflexion flexibility during weight-bearing which is a reflection of Achilles tendon shortening or osseous block in the ankle motrise are the pathogenetic mechanisms of pes cavus in CMT1A (6). Charcot-Marie-Tooth disease should be suspected if a patient has cavus foot and distal symmetric atrophy. Orthotic treatment of the feet can improve the quality of life of patients with CMT by increasing their stability and confidence when walking [10].

Here we present a CMT patient with pes cavus and hammer toes who had been undiagnosed formerly by his medical practitioner and a neurosurgeon.

Case Report

A 16-year-old male was admitted to Physical Medicine and Rehabilitation department in November 2009 with the complaint of progressive gait disturbance and leg weakness and drop left hand for 3 years. At 11 years of age he presented with progressive tingling in his fingers and toes, diminished left hand strength, and difficulty climbing stairs. He had been admitted to a neurosurgery clinic in another centre 4 months ago but any diagnosis had not been yield.

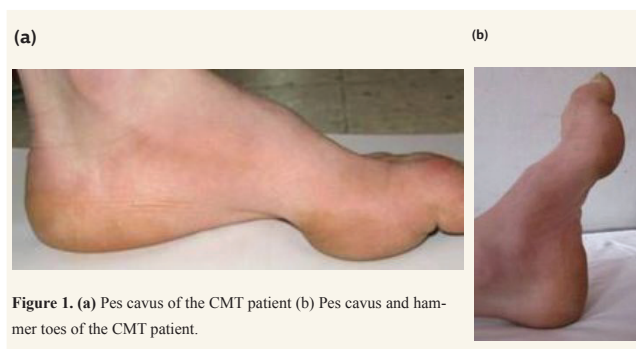


Figure 1. (a) Pes cavus of the CMT patient (b) Pes cavus and hammer toes of the CMT patient.

Examination revealed bilateral pes cavus, hammer toes (Figure 1a, 1b) and poor grip strength, with thinning of the left hand and distal forearm musculature. He had reduced strength in the lower extremities, atrophy of the intrinsic left hand muscles, decreased deep tendon reflexes in the lower extremities, and difficulty walking on his heels. Sensation was diminished in the left hand and feet to temperature, pinprick, and vibration. Tone was normal in the upper extremities but was increased at the ankles. He had difficulty maintaining balance. Upon neurological examination, there was mild weakness (4/5) in knee flexion and extension, and ankle dorsiflexion and planter flexion were 3/5, and left hand dorsiflexion was 1/5. Atrophy was noted in intrinsic feet, left hand and peroneal muscle groups. Nerve conduction velocity measurements disclosed marked slowing in both the left radial and left median and the bilateral tibial nerves. The left radial sensory, left median sensory, left ulnar sensory, and bilateral sural sensory latencies were prolonged, with decreased amplitudes (demyelinating form with low NCVs.). Investigation with electromyography revealed minimal voluntary activity in the foot intrinsec muscles bilaterally and left hand muscles. There was no evidence of conduction block. We diagnosed the patient as HSMN (CMT) according to the clinical and electrophysiological findings.

Discussion

The pes cavus foot appears in childhood as a complex deformity. While the etiology may occasionally be idiopathic, its appearance is often the initial manifestation of a progressive neuromuscular disorder. The deformity presents significant difficulties both the patient and the physiotherapy staff. The symptomatic pes cavus foot can be disabling. The deformity and the pain worsen with time. There are three broad etiologic categories resulting in a pes cavus deformity [11]. The congenital pes cavus category (includes the arthrogryptic foot, the residua of talipes, equinovarus and the idiopathic cavovarus). Posttraumatic pes cavus category that are partially neuromuscular in origin (compartment syndrome or crush injury) and partially structural in origin (fracture malunion or burn contracture). This group is uncommon and difficult to reconstruct. The neuromuscular pes cavus category include poliomyelitis, cerebral palsy, spinal dysraphism, and degenerative diseases of the nervous system such as CMT disease is the most common, it is also the most important, since many cases of pes cavus deformity represent the early signs of a progressive CMT disease. CMT disease encompasses a group of clinically and genetically heterogeneous peripheral neuropathies. This disease affects males more than females by a 5.1:3 ratio [12]. The most commonly used CMT classification combines clinical findings with the pattern of inheritance (autosomal dominant, autosomal recessive, or X-linked) and the electrophysiological findings or those from pathological anatomy (demyelination or axonal degeneration). There are 7 main categories of CMT. Most cases can be included in type CMT 1A (demyelinating forms, with low conduction velocities and autosomal dominant transmission) characterized by progressive foot and ankle weakness, contractures and pes cavus deformity [6,7]. Patients may have diminished deep tendon reflexes, dysphasia and French nerve involvement [13]. There is also a reported 10 percent incidence of scoliosis [14]. The characteristic changes seen in the upper extremities are intrinsic atrophy of the hands and weakness of the radially innervated muscle of the forearms. The foot often has cavus deformities and claw toes. Also, the forefoot has a tendency to be adducted and the heel to be in varus [15]. Since the first and most severely affected function in all cases is ambulation, orthoses are essential [16]. Orthotic treatment of the feet can improve the

quality of life of patients with CMT by increasing their stability and confidence when walking. In the large majority of patients there is no severe weakness in the quadriceps and the glutei muscles, and ambulation is preserved for the whole life even though special footwear and orthotics are necessary to correct footdrop, foot rotation and plantarflexor failure, which are the major problems in CMT1A [17]. A specific design should be made for each patient, in accordance with the morphology and functionality of the foot. However, foot orthoses for patients with CMT should always include a rocker sole in their design. This component should cover the entire forefoot. It should be wide and not very thick [10]. Rotation and walking is corrected by wedges under the foot orthoses and the sole of the shoe with a bit of heel [18]. Surgery is essential for severe foot deformities in CMT.

Exercise intolerance and undue fatigue are common complaints in patients with Charcot-Marie-Tooth (CMT) disease [19]. Conventional rehabilitation programs with interval-training exercise (ITE) improves cardiorespiratory capacities, isokinetic concentric strength, functional ability measurements, fatigue and pain during

training in CMT patients [19]. At present, there is no etiological medical treatment in CMT. High dose Ascorbic acid may repress PMP22 gene expression by acting on intracellular cyclic adenosine monophosphate levels and adenylate cyclase activity and induce remyelination and improve locomotor functioning in CMT1A patients [20].

As a result we conclude that CMT should come into mind in the differential diagnosis of pes cavus and hammer toes.

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